We Claim:

1. A method for determining whether a subject is suffering from Schwachman-Diamond Syndrome (SDS) comprising

obtaining a nucleic acid sample from the subject, and

conducting an assay on the nucleic acid sample to determine the presence or absence of a *SBDS* gene mutation associated with SDS, wherein the presence of a *SBDS* gene mutation associated with SDS in both *SBDS* alleles indicates that the subject suffers from SDS.

2. The method of claim 1 wherein the assay is selected from the group consisting of probe hybridisation, direct sequencing, restriction enzyme fragment analysis and fragment electrophoretic mobility.

3. The method of claim 2 wherein the nucleic acid sample is a DNA sample or an RNA sample and the assay is a direct sequencing assay.

4. The method of claim 3 wherein the nucleic acid sample is a genomic DNA sample and the assay comprises the steps of:

(a) amplifying a target portion of the nucleotide sequence of the genomic DNA;

(b) obtaining the nucleotide sequence of said amplified target portion; and

(c) determining the presence or absence of a *SBDS* gene mutation associated with SDS in said target portion of the nucleotide sequence.

5. The method of claim 3 wherein the nucleic acid sample is an RNA sample and the assay comprises the steps of:

(a) reverse transcribing the RNA sample to produce a corresponding cDNA;

(b) performing at least one polymerase chain reaction with suitable oligonucleotide primers to amplify the *SBDS* cDNA;

(c) obtaining the nucleotide sequence of the amplified *SBDS* cDNA; and

(d) determining the presence or absence of a *SBDS* gene mutation associated with SDS in said nucleotide sequence.

6. The method of claim 4 or 5 wherein the presence or absence of a mutation selected from the group consisting of 24C>A; 97_97insA; 119delG; 131A>G; 183TA>CT; 183TA>CT + 201A>G+258+2T>C; 199A>G; 258+2T>C; 258+1G>C; 260T>G; 291_293delTAAinsAGTTCAAGTATC; 377G>C; 505C>T+651C>T, 183_184TA CT; 183_184TA CT+258+2T C; 258+2T C; 24C A; 96-97insA; 119delG; 131A G; 199A G; 258+1G C; 260T G; 291-293delTAAinsAGTTCAAGTATC; 377G C; 505C T; 56G A; 93C G; 97A G; 101A T; 123delC; 279_284delTCAACT; 296_299delAAGA; 354A C; 428C T+443A G; 458A G; 460-1G A; 506G C; and 624+1G C is determined.

7. The method of claim 4 or 6 wherein the target portion of the nucleotide sequence is amplified using a primer pair selected from the group consisting of:

(a) Primer A and Primer B;

(b) Primer E and Primer F;

(c) Primer G and Primer H;

(d) Primer SDCR9x4seqB and Primer J;

(e) Primer SDCR9x5Fseq and Primer L;

(f) Primer Q and Primer B;

(g) Primer I and Primer J;

(h) Primer K and Primer L; and

(i) Primer SDCR9prom1RA and Primer SDCR9prom6FA.

8. The method of claim 2 wherein the nucleic acid sample is a DNA sample and the assay is a restriction enzyme fragment analysis.

9. The method of claim 8 wherein the assay comprises the steps of:

(a) digesting the DNA with a restriction enzyme to give restriction fragments;

(b) separating the restriction fragments by agarose gel electrophoresis; and

(c) detecting the separated fragments by hybridisation of the fragments to a detectably labelled nucleotide probe specific for *SBDS*.

10. The method of claim 9 wherein the restriction enzyme is at least one of Cac81 and Bsu361.

11. The method of any one of claims 1 to 10 wherein the subject is a human subject.

12. A method for determining whether a subject is an SDS carrier comprising

obtaining a nucleic acid sample from the subject, and

conducting an assay on the nucleic acid sample to determine the presence or absence of a *SBDS* gene mutation associated with SDS, wherein the presence of a *SBDS* gene mutation associated with SDS in one *SBDS* allele indicates that the subject is an SDS carrier.

13. The method of claim 12 wherein the assay is selected from the group consisting of probe hybridisation, direct sequencing, restriction enzyme fragment analysis and fragment electrophoretic mobility.

14. The method of claim 13 wherein the nucleic acid sample is a DNA sample or an RNA sample and the assay is a direct sequencing assay.

15. The method of claim 14 wherein the nucleic acid sample is a genomic DNA sample and the assay comprises the steps of:

(a) amplifying a target portion of the nucleotide sequence of the genomic DNA;

(b) obtaining the nucleotide sequence of said amplified target portion; and

(c) determining the presence or absence of a *SBDS* gene mutation associated with SDS in said target portion of the nucleotide sequence.

16. The method of claim 14 wherein the nucleic acid sample is an RNA sample and the assay comprises the steps of:

(a) reverse transcribing the RNA sample to produce a corresponding cDNA;

(b) performing at least one polymerase chain reaction with suitable oligonucleotide primers to amplify the *SBDS* cDNA;

(c) obtaining the nucleotide sequence of the amplified *SBDS* cDNA; and;

(d) determining the presence or absence of a *SBDS* gene mutation associated with SDS in said nucleotide sequence.

17. The method of claim 15 or 16 wherein the presence or absence of a mutation selected from the group consisting of 24C>A; 97_97insA; 119delG; 131A>G; 183TA>CT; 183TA>CT + 201A>G+258+2T>C; 199A>G; 258=2T>C; 258+1G>C; 260T>G; 291_293delTAAinsAGTTCAAGTATC; 377G>C; 505C>T+651C>T; 183_184TA CT; 183_184TA CT+258+2T C; 258+2T C; 24C A; 96-97insA; 119delG; 131A G; 199A G; 258+1G C; 260T G; 291-293delTAAinsAGTTCAAGTATC; 377G C; 505C T; 56G A; 93C G; 97A G; 101A T; 123delC; 279_284delTCAACT; 296_299delAAGA; 354A C; 428C T+443A G; 458A G; 460-1G A; 506G C; and 624+1G C is determined.

18. The method of claim 15 or 16 wherein the target portion of the nucleotide sequence is amplified using a primer pair selected from the group consisting of:

(a) Primer A and Primer B;
(b) Primer E and Primer F;
(c) Primer G and Primer H;
(d) Primer SDCR9x4seqB and Primer J;
(e) Primer SDCR9x5Fseq and Primer L;
(f) Primer Q and Primer B;
(g) Primer I and Primer J;
(h) Primer K and Primer L; and
(i) Primer SDCR9prom1RA and Primer SDCR9prom6FA.

19. The method of claim 13 wherein the nucleic acid sample is a DNA sample and the assay is a restriction enzyme fragment analysis.

20. The method of claim 19 wherein the assay comprises the steps of:

(a) digesting the DNA with a restriction enzyme to give restriction fragments;
(b) separating the restriction fragments by agarose gel electrophoresis; and
(c) detecting the separated fragments by hybridisation of the fragments to a detectably labelled nucleotide probe specific for *SBDS*.

21. The method of claim 20 wherein the restriction enzyme is *Nde* 1.

22. The method of any one of claims 12 to 21 wherein the subject is a human subject.

23. A method for determining whether a subject is suffering from Shwachman-Diamond Syndrome (SDS) comprising

obtaining a tissue sample from the subject, and

conducting an assay on the tissue sample to determine the level of SBDS protein in the sample, wherein a reduced level of SBDS protein in the sample relative to a control sample indicates that the subject suffers from SDS.

24. The method of claim 23 wherein the tissue sample is selected from the group consisting of blood, buccal smear or bone marrow aspirate.

25. A method for determining whether a subject is at risk for developing acute myelogenous leukaemia (AML) comprising

obtaining a nucleic acid sample from the subject, and

conducting an assay on the nucleic acid sample to determine the presence or absence of a *SBDS* gene mutation associated with SDS, wherein the presence of a *SBDS* gene mutation associated with SDS indicates that the subject is at risk for development of AML.

26. A method for treating a subject suffering from SDS comprising administering to the subject a therapeutically effective amount of a substantially purified SBDS protein or of an isolated nucleotide sequence encoding an SBDS protein.

27. The method of claim 26 wherein a sample of bone marrow cells is obtained from the subject and the bone marrow cells are transfected with a nucleotide sequence encoding an SBDS protein and re-introduced into the subject.

28. The method of claim 26 or 27 wherein the nucleotide sequence encodes a protein of amino acid sequence SEQ ID NO:2.

29. The method of claim 26 or 27 wherein the nucleotide sequence is the sequence of SEQ ID NO:1.

30. The method of claim 26 wherein the substantially purified SBDS protein has the amino acid sequence of SEQ ID NO:2.

31. An isolated nucleic acid molecule encoding an SBDS protein.

32. The nucleic acid molecule of claim 31 wherein the protein is a human SBDS protein.

33. The nucleic acid molecule of claim 32 comprising a nucleotide sequence selected from the group consisting of:

(a) the nucleotide sequence of SEQ ID NO:1;

(b) a nucleotide sequence encoding the amino acid sequence of SEQ ID NO:2;

(c) a nucleotide sequence which is a complement of a nucleotide sequence of (a) or (b); and

(d) a nucleotide sequence which hybridises under stringent conditions to a nucleotide sequence of (a) or (b).

34. The nucleic acid molecule of claim 31 wherein the protein is a murine SBDS protein.

35. The nucleic acid molecule of claim 34 comprising a nucleotide sequence which encodes the amino acid sequence of SEQ ID NO:29 or comprising the nucleotide sequence of SEG ID NO:29.

36. The nucleic acid molecule of claim 34 wherein the nucleotide sequence has at least one mutation selected from the group consisting of 24C>A; 97_97insA; 119delG; 131A>G; 183TA>CT; 183TA>CT + 201A>G+258+2T>C; 199A>G; 258+2T>C; 258+1G>C; 260T>G; 291_293delTAAinsAGTTCAAGTATC; 377G>C; 505C>T+651C>T, 183_184TA CT; 183_184TA CT+258+2T C; 258+2T C; 24C A; 96-97insA;

119delG; 131A G; 199A G; 258+1G C; 260T G; 291-293delTAAinsAGTTCAAGTATC; 377G C; 505C T; 56G A; 93C G; 97A G; 101A T; 123delC; 279_284delTCAACT; 296_299delAAGA; 354A C; 428C T+443A G; 458A G; 460-1G A; 506G C; and 624+1G C is determined.

37. The nucleic acid molecule of any one of claims 31 to 35 wherein the molecule is a DNA molecule.

38. The nucleic acid molecule of any one of claims 31 to 35 wherein the molecule is an RNA molecule.

39. A recombinant vector comprising the isolated nucleic acid molecule of any one of claims 31 to 38.

40. A host cell comprising the vector of claim 39.

41. An isolated nucleic acid molecule comprising at least about 10, 20, 30, 50, 75 or 100 consecutive nucleotides of SEQ ID NO:1 or 29.

42. A substantially purified SBDS protein.

43. The protein of claim 42 comprising an amino acid sequence selected from the group consisting of:

(a) the amino acid sequence of SEQ ID NO:2;

(b) the amino acid sequence of SEQ ID NO:29.

44. An antibody which binds specifically to an epitope of an SDS protein.

45. The antibody of claim 44 wherein the antibody binds specifically to an SBDS protein having at least 89% amino acid identity with a protein comprising the amino acid sequence of SEQ ID NO:2 or SEQ ID NO:29.

46. A hybridoma cell line which produces an antibody in accordance with claim 44 or 45.

47. A method for preparing an SBDS protein comprising expressing the nucleotide sequence of any one of claims 31 to 38 in a suitable expression system and collecting the expressed protein.

48. A nucleotide sequence selected from the group consisting of:

(a) 5'-GCGTAAAAAGCCACAATAC-3' (SEQ ID NO:3);
(b) 5'-CTATGACAGTATTCGTAAGACTAGG-3' (SEQ ID NO:4);
(c) 5'-GGGGATTTGTTGTGTCTTG-3' (SEQ ID NO:5);
(d) 5'-CTTTCCTCCAGAAAAACAGC-3' (SEQ ID NO:6);
(e) 5'-AAATGGTAAGGCAAATACGG-3' (SEQ ID NO:7);
(f) 5'-ACCAAGTTCTTTATTATTAGAAGTGAC-3' (SEQ ID NO:8);
(g) 5'-GCTCAAACCATTACTTACATATTGA-3' (SEQ ID NO:9);
(h) 5'-CACTTGCTTCCATGCAGA-3' (SEQ ID NO:10);
(i) 5'-AAAGGGTCATTTTAACACTTC-3' (SEQ ID NO:11);
(j) 5'-GAAAATATCTGACGTTTACAACA-3' (SEQ ID NO:12);
(k) 5'-TCCACTGTAGATGTGAACTAACTC-3' (SEQ ID NO:13);
(l) 5'-CACTCTGGACTTTGCATCTT-3' (SEQ ID NO:14);
(m) 5'-GCTTCTGCTCCACCTGAC-3' (SEQ ID NO:15);
(n) 5'AGCTATGCTGCAGCTGTTAC-3' (SEQ ID NO:16);
(o) 5'-ATGCATGTCCAAGTTTCAAG-3' (SEQ ID NO:17);
(p) 5'-TCCATGGCTATATTTTGATGA-3' (SEQ ID NO:18);
(q) 5'-TAAGCCTGCCAGACACAC-3' (SEQ ID NO:19);
(r) 5'-CACTCTGGACTTTGCATCTT-3' (SEQ ID NO:20);
(s) 5'-TGTTGGTTTTCACCGAATA-3' (SEQ ID NO:21);
(t) 5'-AGATAAAGAAAGACACACACAACT-3' (SEQ ID NO:22);
(u) 5'-GAAATCGCCTGCTACAAA-3' (SEQ ID NO:23);
(v) 5'-TCAGCTTCTTGCCTTCAT-3' (SEQ ID NO:24);
(w) 5'-TAAGTAAGCCTGCCAGACA-3' (SEQ ID NO:25);
(x) 5'-CATCAAGGTCTTTTTCCAAG-3' (SEQ ID NO:26);

(y) 5'-CCTGTCTCTGCCCAAGTC-3' (SEQ ID NO:27); and

(z) 5'-AGGGAACATTTTCAAAACTCA-3' (SEQ ID NO:28).

49. A transgenic non-human mammal having within its genome an SBDS gene with at least one mutation associated with SDS.

50. The mammal of claim 49 wherein the mammal is selected from the group consisting of mice, rats, rabbits, sheep, goats and non-human primates.

51. The mammal of claim 49 wherein the mammal is a mouse.

52. A kit comprising at least one pair of primers suitable for amplification of at least a portion of an SBDS gene.